Risk Factors for Late Onset Hearing Loss: Features Associated with Syndromes

Left undetected, hearing loss in infants can negatively impact speech and language acquisition, academic achievement, and social and emotional development. If detected early, however, morbidity can be diminished and even eliminated through early intervention services. This fact sheet reviews some of the defining characteristics of a number of syndromes associated with hearing loss.

Background

There are over 400 genetic syndromes that are associated with hearing loss; the inheritance pattern, prevalence, and description of a few of the more common syndromes are listed below. Approximately 50% of cases of prelingual deafness are attributed to genetic factors; about 30% of these are thought to be part of a syndrome. Genetic syndromes may be autosomal dominant (in which a single mutation in a gene on one of the autosomes [non-sex chromosomes] from either parent can result in the syndrome), autosomal recessive (a pair of matching autosomal genetic mutations, one from each parent, results in the syndrome), or X-linked (the mutation in the gene responsible for the syndrome is located on the X chromosome). A three-generation family history can aid in the diagnosis of the specific cause of the hearing loss and can aid in determining the recurrence risk.

Autosomal Dominant Syndromes

- *Waardenburg Syndrome*
  Prevalence: 1/20,000 to 1/40,000 births
  Description: congenital sensorineural hearing loss, hair hypopigmentation (usually white forelock, eyebrows, and/or eyelashes), pigmentation abnormality of the iris, and dystopia canthorum (a widened bridge of the nose because of lateral placement of inner canthus of the eyes).

- *Branchio-Oto-Renal (BOR) Syndrome*
  Prevalence: unknown (estimated to be between 1/40,000 and 1/700,000 in general population)
  Description: diagnosis without family history based on the presence of any three of the following: mild to profound conductive, sensorineural, or mixed hearing loss (due to abnormalities of the pinnae, external auditory canal, middle ear, or inner ear), pre-auricular pits or tags, “lop-ear” deformity, branchial fistulae (a passage from the side of the neck and leading into the pharynx), and renal anomalies.

- *CHARGE Syndrome*
  Prevalence: 1/12,000 births
  Description: **C**oloboma of the eye (a congenital cleft, hole, or split in the iris), **H**eart anomaly, choanal **A**tresia (blockage of the nasal cavity due to bony or membranous tissue), **R**etardation of mental & somatic development, microphallus (**G**enital and urinary difficulties), and **E**ar anomalies and/or deafness (mixed sensorineural and conductive).

- *Stickler Syndrome*
  Prevalence: estimated to be 1/7,500 births
  Description: flat face with depressed nasal bridge, midfacial hypoplasia (underdeveloped midfacial regions), high myopia (nearsightedness), arthritis after 30 years of age, hypotonia, hyperextensible joints, prominence of large joints may be present at birth, variable (sometimes progressive) hearing impairment, Pierre Robin sequence (small or set-back lower jaw, displaced tongue, cleft palate), and mitral valve prolapse.

- *Neurofibromatosis Type II*
  Prevalence: 1/40,500 to 1/87,410 births
  Description: schwannoma (benign tumor of nerve fibers), spinal tumors, meningioma (a tumor that grows from the meninges, the membranes that cover the brain and spinal cord), juvenile posterior subcapsular cataracts, mono/poly neuropathy (a disorder of a single or multiple peripheral nerves), hearing loss, focal weakness (occurring in one area of the body), tinnitus (ringing in the ears), balance dysfunction, seizure, and bilateral acoustic neuromas developing usually in the second or third decade.

Autosomal Dominant Syndromes — continued

- *Treacher Collins Syndrome*
  Prevalence: 1/10,000 to 1/50,000 live births
  Description: midface hypoplasia (underdeveloped midfacial regions), micrognathia (small lower jaw) and retrognathia (lower jaw is set back from the upper jaw), external auditory canal defects, conductive hearing loss, coloboma of the lower eyelid (a congenital cleft, hole, or split), partial or total absence of lower eyelashes, and preauricular hair displacement.

Autosomal Recessive Syndromes

- *Usher Syndrome*
  Prevalence in the US: 1/23,000
  Description: congenital bilateral mild to severe hearing loss or progressive sensorineural hearing loss, vestibular areflexia (absence of neurological reflexes in the vestibular area), and retinitis pigmentosa (an eye disorder that affects night and peripheral vision).

- *Pendred Syndrome*
  Prevalence: unknown
  Description: hearing loss that is usually congenital (sometimes late onset and progressive), vestibular dysfunction, temporal bone anomalies, and goiter.

- *Jervell & Lange-Nielsen Syndrome (Long QT Syndrome)*
  Prevalence: 1.6-6/million in general population
  Description: profound congenital sensorineural deafness, prolonged QTc interval (heart takes longer to repolarize after each beat), often presents as a deaf child with syncopal episodes (fainting) during stress, exercise, or fright. May have a family history of sudden death or seizure (before age 40).

X-Linked Syndromes

- *Alport Syndrome*
  Prevalence: 1/50,000 live births
  About 80% of Alport syndrome is X-linked, about 15% is autosomal recessive, and about 5% is autosomal dominant.
  Description: microhematuria (the presence of microscopic amounts of blood in the urine), ocular lesions, anterior lenticonus (slowly progressive deterioration of vision), maculopathy, hearing loss is never congenital but can be detected in late childhood or early adulthood.

References

1. Gene Reviews, www.genetests.com (accessed March 7, 2006)
2. Online Mendelian Inheritance in Man, http://www3.ncbi.nlm.nih.gov/entrez/dispomim.cgi?id=214800 (accessed March 7, 2006)

For more information about infant hearing loss, please visit our website: www.doh.wa.gov/ehddi.

Revised 3/28/2006

Contact us at:

Washington State Department of Health
Early Hearing Loss, Detection, Diagnosis, and Intervention (EHDDI) Program
1610 NE 150th Street, K17-9, Shoreline, WA 98155-0729
Phone: (206) 418-5613,
E-mail: ehddi2@doh.wa.gov

EHDDI

Washington State Department of Health